WHAT IS CLAIMED IS:

1. An isolated nucleic acid molecule comprising:

(a) a nucleic acid sequence encoding a human aspartoacylase polypeptide;

(b) a nucleic acid sequence complementary to nucleic acid sequence (a); or

(c) a nucleic acid sequence at least 16 nucleotides in length capable of hybridizing, under stringent hybridization conditions, with one of said nucleic acid molecules (a) or (b).

2. A nucleic acid molecule of claim 1(a), comprising:

(i) a DNA molecule having the DNA sequence of Fig. 1 from DNA position +1 to +891;

(ii) a DNA molecule encoding a normal human aspartoacylase polypeptide having the amino acid sequence of Fig. 1 from amino acid position +1 to +313;

(iii) a DNA molecule having a sequence of a fragment of the DNA sequence of Fig. 1, or a sequence complementary thereto, and including at least 16 sequential nucleotides;

(iv) a DNA molecule having a sequence of a fragment of the DNA sequence of Fig. 1 and including at least 16 sequential nucleotides, and which encodes a fragment of the amino acid sequence of Fig. 1; or

(v) a DNA molecule encoding an epitope of the amino acid sequence of Fig. 1 between positions +1 to +313, and encoded by at least 18 sequential nucleotides.

3. A nucleic acid molecule of claim 1(a), comprising

(i') a DNA molecule having the DNA sequence of Fig. 1 from DNA position +1 to +891;

(ii') a DNA molecule encoding a normal human aspartoacylase polypeptide having the amino acid sequence of Fig. 1 from amino acid position +1 to +313;

(iii') a DNA molecule having a sequence of a fragment of the DNA sequence of Fig. 1, or a sequence complementary thereto, and including at least 16 sequential nucleotides;

(iv') a DNA molecule having a sequence of a fragment of the DNA sequence of Fig. 1 and including at least 16 sequential nucleotides, and which encodes a fragment of the amino acid sequence of Fig. 1; or

(v') a DNA molecule encoding an epitope of the amino acid sequence of Fig. 1 between positions +1 to +313, and encoded by at least 18 sequential nucleotides,

except that each of said DNA molecules (i'), (ii'), (iii'), (iv') or (v') is modified in at least one nucleotide position as compared with the sequence of Fig. 1, corresponding to the sequence of a naturally-occurring allele of human aspartoacylase having an altered biological activity.

4. An isolated DNA molecule of claim 1, comprising a DNA sequence encoding a human aspartoacylase polypeptide.

5. An isolated DNA molecule of claim 4, wherein the DNA codes for a normal human aspartoacylase polypeptide.

6. An isolated DNA molecule of claim 4, wherein the DNA codes for a naturally-occurring allele or a mutant of human aspartoacylase

polypeptide having an altered biological activity as compared with a normal aspartoacylase polypeptide.

7. An isolated DNA molecule of claim 6, wherein the DNA codes for an aspartoacylase polypeptide which, if expressed in cells of the human body, is associated with altered cellular function which correlates with Canavan disease.

8. An isolated nucleic acid primer or probe molecule comprising a DNA or RNA nucleotide sequence corresponding to the sequence (iii), (iv) or (v), or a sequence complementary thereto.

9. An isolated nucleic acid primer or probe molecule comprising a DNA or RNA nucleotide sequence corresponding to the sequence (iii'), (iv') or (v'), or a sequence complementary thereto.

10. A nucleic acid molecule of claim 8, having the sequence
CTTCTGAATTGCAGAAATCA (HASP9) or
GTAAGACACCGTGTAAGATG (HASPC7).

11. A nucleic acid molecule of claim 9, having the sequence
F→CCGGGATGAAAATGGAGAA (HASP14F) or
R→ACCGTGTAAGATGTAAGC (HASPC7R),
wherein F and R are M13 universal and/or reverse primer tags.

12. A DNA molecule of claim 5, having the DNA sequence of Fig. 1 from DNA position +1 to +891.

13. A DNA molecule of claim 7, having the DNA sequence of Fig. 1 from DNA position +1 to +891, except that the adenine nucleotide at position 854 is replaced by a cytosine nucleotide.

14. A recombinant vector comprising a DNA molecule of claim 1.

15. A vector of claim 14, wherein said DNA molecule is operably linked to an expression control sequence suitable for expression of said DNA sequence in a host cell.

16. A host cell transformed with a vector of claim 15.

17. A host cell of claim 16, selected from a strain of *E. coli*, *Pseudomonas*, *Bacillus subtilis*, *Bacillus stearothermophilus*, or other bacilli; other bacteria; yeast; other fungi; insect cells; plant cells; or murine, bovine, porcine, human or other mammalian cells.

18. A method of producing a normal aspartoacylase polypeptide, comprising

(a) culturing a host cell transformed with a vector of claim 15 containing a DNA coding for a normal aspartoacylase polypeptide in a cell culture medium under conditions whereby the aspartoacylase polypeptide is expressed, and

(b) isolating the thus-produced normal aspartoacylase polypeptide.

19. A method of producing a mutant aspartoacylase polypeptide having an altered biological activity as compared with a normal aspartoacylase polypeptide, comprising

(a) culturing a host cell transformed with a vector of claim 15 containing a DNA coding for a mutant aspartoacylase polypeptide in a cell culture medium under conditions whereby the aspartoacylase polypeptide is expressed, and

(b) isolating the thus-produced mutant aspartoacylase polypeptide.

20. An isolated normal aspartoacylase polypeptide capable of hydrolyzing N-acetyl-aspartic acid to aspartate and acetate.

21. A normal aspartoacylase polypeptide of claim 20, having the amino acid sequence of Fig. 1 from amino acid position +1 to +313.

22. A mutant aspartoacylase polypeptide having either an altered ability of hydrolyze N-acetyl-aspartic acid to aspartate and acetate or incapable of hydrolyzing N-acetyl-aspartic acid to aspartate and acetate.

23. A mutant aspartoacylase polypeptide of claim 22, having the amino acid sequence of Fig. 1 from amino acid position +1 to +313, except that the glutamic acid at amino acid position 285 is substituted by another amino acid or is deleted.

24. A mutant aspartoacylase of claim 23, wherein the glutamic acid at amino acid position 285 is substituted by alanine.

25. An isolated naturally occurring gene, comprising a DNA sequence of claim 1 linked to naturally occurring control sequences.

26. An isolated gene of claim 25, encoding and capable of expressing a normal aspartoacylase polypeptide.

27. An isolated gene of claim 25, which is not capable of expressing a normal aspartoacylase polypeptide.

28. An isolated gene of claim 27, encoding and capable of expressing a mutant aspartoacylase polypeptide having an altered ability of hydrolyze N-acetyl-aspartic acid to aspartate and acetate or incapable of hydrolyzing N-acetyl-aspartic acid to aspartate and acetate.

29. An isolated gene of claim 28, which, if expressed in cells of the human body, is associated with altered cellular function which correlates with Canavan disease.

30. An isolated gene of claim 27, encoding a mutant expression control sequence which is not capable of expression of aspartoacylase.

31. A method of screening a subject to determine if said subject is a Canavan carrier or a Canavan patient, comprising

(a) providing a biological sample of the subject to be screened; and

(b) submitting the sample to an assay for detecting in the biological sample the presence of a normal aspartoacylase gene, a mutant aspartoacylase gene, normal aspartoacylase polypeptide, mutant aspartoacylase polypeptide, or mixtures thereof.

32. A method of claim 31, wherein the biological sample includes at least part of the genome of the subject and the assay comprises a hybridization assay.

33. A method of claim 31, wherein the assay comprises at least one labeled nucleotide probe.

34. A method of claim 33, wherein the assay comprises a labeled nucleotide probe comprising a DNA molecule having a sequence of a fragment of the DNA sequence of Fig. 1, or a sequence complementary thereto, and including at least 16 sequential nucleotides.

35. A method of claim 33, wherein the probe is selected from the sequences

CTTCTGAATTGCAGAAATCA (HASP9),
GTAAGACACCGTGTAAGATG (HASPC7),
F→CCGGGATGAAAATGGAGAA (HASP14F) or
R→ACCGTGTAAGATGTAAGC (HASPC7R),

wherein F and R are M13 universal and/or reverse primer tags.

36. A method of claim 31, wherein the biological sample includes an aspartoacylase polypeptide of the subject and the assay is an immunoassay and comprises an antibody.

37. A method of claim 36, wherein the antibody is specific for a normal aspartoacylase polypeptide.

38. A method of claim 36, wherein the antibody is specific for a mutant aspartoacylase polypeptide.

39. A method of claim 36, wherein the assay is a radioimmunoassay.

40. A method of claim 36, wherein the antibody is a monoclonal antibody.

41. A process for screening a potential Canavan disease carrier or patient for the presence of an identified mutation in an aspartoacylase gene, comprising

(a) isolating genomic DNA from said potential Canavan disease carrier or patient,

(b) hybridizing a DNA probe onto said isolated genomic DNA, said DNA probe spanning said mutation in said aspartoacylase gene, wherein said DNA probe is capable of detecting said mutation,

(c) treating said genomic DNA to determine the presence or absence of said DNA probe and thereby indicating the presence or absence of said aspartoacylase mutation.

42. A process for screening a potential Canavan disease carrier or patient for the presence of an identified mutation in an aspartoacylase gene, comprising

(a) isolating genomic DNA from said potential Canavan disease carrier or patient,

(b) determining the presence or absence of a restriction endonuclease site in the gene, the presence or absence of which thereby indicates the presence or absence of said aspartoacylase mutation.

43. A process for screening a potential Canavan disease carrier or patient for the presence of an identified mutation in an aspartoacylase gene, comprising

(a) isolating genomic DNA from said potential Canavan disease carrier or patient,

(b) determining the mobility of heteroduplex PCR products in polyacrylamide gels, the mobility of which thereby indicates the presence or absence of said aspartoacylase mutation.

44. A kit for assaying for the presence of an aspartoacylase gene by immunoassay, comprising:

(a) an antibody which specifically binds to a gene product of the aspartoacylase gene;

(b) a reagent means for detecting binding of the antibody to the gene product; wherein

the antibody and reagent means are each present in amounts effective to perform the immunoassay.

45. A kit of claim 44, wherein said reagent means for detecting binding is selected from the group consisting of fluorescence detection, radioactive decay detection, enzyme activity detection or colorimetric detection.

46. A kit of claim 45, wherein said aspartoacylase gene is the normal aspartoacylase gene.

47. A kit of claim 45, wherein said aspartoacylase gene is the mutant aspartoacylase gene.

48. A kit for assaying for the presence of an aspartoacylase gene by hybridization assay, comprising:

(a) an oligonucleotide probe which specifically binds to the aspartoacylase gene;

(b) a reagent means for detecting binding of the hybridization of the oligonucleotide probe to the gene; wherein

the probe and reagent means are each present in amounts effective to perform the hybridization assay.

49. A kit of claim 48, wherein said aspartoacylase gene is the normal aspartoacylase gene.

50. A kit of claim 48, wherein said aspartoacylase gene is the mutant aspartoacylase gene.

51. An immunologically active anti-aspartoacylase polyclonal or monoclonal antibody specific for an aspartoacylase polypeptide of claim 20.

52. An immunologically active anti-aspartoacylase polyclonal or monoclonal antibody specific for an aspartoacylase polypeptide of claim 22.

53. A hybridoma producing a monoclonal antibody specific for an aspartoacylase polypeptide of claim 20.

54. A hybridoma producing a monoclonal antibody specific for an aspartoacylase polypeptide of claim 22.

55. A method of treatment for Canavan disease in a patient, comprising administering to the patient a therapeutically effective amount of the polypeptide of claim 20.

56. A method of gene therapy for Canavan disease in a patient, comprising delivering to a cell of said patient a DNA molecule of claim 1.

57. A method of claim 56, wherein the delivery step further comprises the step of providing a vehicle for delivery.

58. A method of claim 57, wherein the vehicle is a recombinant vector.

59. An animal comprising a heterologous cell system, comprising a recombinant cloning vector of claim 14, which induces Canavan disease symptoms in said animal.

60. An animal of claim 59 wherein said animal is a mammal.

61. An animal of claim 60, which is a rodent.

62. An animal of claim 61, wherein said rodent is a mouse.

63. A transgenic mouse exhibiting Canavan disease symptoms.

64. A kit of claim 48, further comprising oligonucleotide probes specific for assaying for the presence or absence of one or more additional genes, mutant or normal, and, optionally, other regent means specific for said additional genes, which additional genes code for proteins involved in other genetic diseases.

65. A kit of claim 64, wherein the second genetic disease is Tay-Sachs disease and/or Goucher disease.